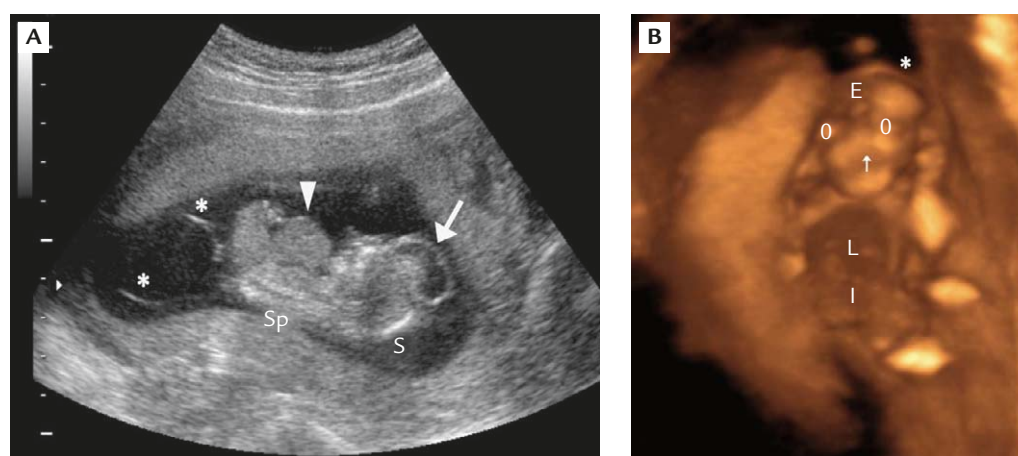


# TYPICAL BODY WALL DEFECT ASSOCIATED WITH CRANIOFACIAL ANOMALIES AND AMNIOTIC BANDS DIAGNOSED IN EARLY PREGNANCY

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**Figure 1.** At 13 weeks of gestation, (A) two-dimensional ultrasonography shows acrania, exencephaly (arrow), extracorporeal visceral organs (arrowhead), and amniotic bands (asterisks) but no fetoplacental attachment; and (B) three-dimensional ultrasonography shows median facial clefting (arrow), acrania, exencephaly, body wall defect with extracorporeal liver and intestines, and amniotic bands (asterisk). S=skull; Sp=spine; E=exencephaly; O=orbit; L=liver; I=intestines.

A 30-year-old primigravid, a woman was referred to the hospital at 13 weeks of gestation because of abnormal sonographic findings indicating fetal malformations. The woman reported no recent infections, invasive genetic diagnosis, maternal trauma or other illness, no prenatal exposure to teratogenic agents, and no family history of congenital malformations or hereditary collagen disorders. Two-dimensional ultrasonography demonstrated acrania with an absence of flat skull bones, disorganized cerebral hemispheres, body wall defect with extracorporeal visceral organs, and amniotic bands adherent to the fetus (Figure 1A). Three-dimensional ultrasonography demonstrated facial clefting, hypertelorism, amniotic bands, skull defects, exencephaly, and

abdominal wall defect with extracorporeal liver and intestines (Figure 1B). There were no limb deficiency, no cranioplacental attachment, and no abdominoplacental attachment. The amniotic fluid volume was normal. A diagnosis of amniotic band syndrome (ABS) was made. The pregnancy was terminated subsequently. A male fetus weighing 24 g was delivered, with a karyotype of 46,XY. The multiple anomalies included acrania, exencephaly, median facial clefting, nasal deformity, aberrant bands over the face, hypertelorism, body wall defects with extracorporeal liver and intestines, and constrictive amniotic bands over the fingers and placental surface (Figure 2). There was no deficiency of the limbs or fetoplacental attachment. The male external genitalia, the anal opening, and the umbilical cord were normal.

We have presented the earliest prenatal sonographic demonstration of ABS with both craniofacial and abdominal wall defects but without fetoplacental attachment. In the present case, prenatal ultrasonography

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**Figure 2.** The fetus at birth.

helps with prenatal differential diagnosis of ABS from limb-body wall complex (LBWC), omphalocele, and classic neural tube defects in early gestation.

Toprin [1] suggested that in ABS, multiple fetal anomalies result from a rupture of the amniotic sac. The constrictive amniotic bands following a primary amniotic rupture can interrupt the normal embryonic craniofacial and body development, leading to severe malformations and disruptions of the cranio-face and body wall [2]. LBWC with craniofacial defects frequently shows severe anomalies of the upper limbs, craniofacial defects, constrictive amniotic bands, and cranioplacental attachment, whereas LBWC without craniofacial defects usually presents with major anomalies of the lower limbs, abnormal genitalia, anal atresia, renal defects, abdominoplacental attachment, and umbilical cord abnormalities [3]. Russo et al [4] and

Russo and Vecchione [5] suggested that LBWC with craniofacial defects is caused by an early vascular disruption, and LBWC without craniofacial defects is related to a defective lateral and caudal folding process of the embryonic disk. Most cases of ABS are sporadic and have no risk of recurrence. However, cases with classic neural tube defects due to multifactorial etiology carry a 2–5% risk of recurrence [6], and 11–66.7% of the fetuses with omphalocele have cytogenetic abnormalities [7]. Prenatal diagnosis of acrania warrants a careful survey of the fetal limbs, body wall, internal organs, face and placenta, and for the presence of amniotic bands. A correct perinatal diagnosis of ABS is important for genetic counseling and estimation of the recurrent risk.

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